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Promotion and protection of all human rights, civil, political, economic, social and cultural rights, including the right to development

Joint written statement* submitted by Thalassaemia International Federation Limited, Agrenska Foundation (Grosshandlare Axel H. Agrens Donationsfond), International Federation for Hydrocephalus and Spina Bifida - IF, non-governmental organizations in special consultative status

The Secretary-General has received the following written statement which is circulated in accordance with Economic and Social Council resolution 1996/31.

[25 May 2018]

* This written statement is issued, unedited, in the language(s) received from the submitting non-governmental organization(s).

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The Human Rights of People living with a Rare Disease*

As NGOSs with ECOSOC Consultative Status, Thalassaemia International Federation Limited, Agrenska Foundation (Grosshandlare Axel H. Agrens Donationsfond) and International Federation for Hydrocephalus and Spina Bifida – IF, fully support the messages laid out in the contribution made by the **NGO Committee for Rare Diseases1** in the context of the open consultation by the Office of the High Commissioner for Human Rights (OHCHR) for the "Report on the contributions of the right to health framework to the effective implementation and achievement of the health-related Sustainable Development Goals"2. The submission, entitled 'Right to Health in Rare Diseases', puts forward the following messages:

Resolution 35/23, adopted by the Human Rights Council on 23 June 2017, enshrines ***"the right of everyone to the enjoyment of the highest attainable standard of physical and mental health"*** not only as a central component of the 2030 Agenda for Sustainable Development, bu.. also as an essential instrument for its actual implementation.

Health intersects in a structural way several of the Sustainable Development Goals, well beyond SDG 3 only. **The highest attainable standard of physical and mental health represents the foundational bedrock without which an individual cannot be genuinely enabled to set forth on any pursuit of life**, be it towards personal, intellectual, economic or social fulfillment. Only by upholding human rights "*to health and through health*", will the delivery of the SDGs indeed "*leave no one behind*".3

Such considerations are of particular relevance in the field of rare diseases, which gather well over 6,000 conditions, often of genetic origin but also correlated to other types of infectious or environmental agents, and which affect small to ultra-small populations of patients (typically fewer than 1 in 2,000 individuals). Most rare diseases are chronic, progressive, degenerative, disabling and frequently life-threatening. Although each disease taken in isolation is rare, the estimated 300 million affected people worldwide **share many of the same challenges, across diseases and across borders.**

Several of the challenges faced by persons with rare diseases intersect the main pillars of the "right to health" framework:

- *Accessibility* is very often one of the most severe limitations faced by persons with rare diseases. Accessibility bottlenecks are often experienced in the field of:
    - proper and timely **access to treatments and health services and care**4
    - **access to understandable information** about an individual's own condition and to an accurate and timely diagnosis5
    - **access to infrastructure**, and even more broadly of access to equal opportunities (e.g. education, career, reproductive health, etc.).

1 For further information, please refer to www.ngocommitteerarediseases.org. Please see www.ngocommitteerarediseases.org/members/ and www.ngocommitteerarediseases.org/members/executive-board/ for details of the Committee's members and of our Inception Executive Board formed 11 Nov. 2017.

2 http://www.ohchr.org/EN/Issues/ESCR/Pages/HealthFramework.aspx

3 Report of the High-Level Working Group on the Health and Human Rights of Women, Children and Adolescents (2017) "*Leading the Realization of Human Rights to Health and through Health*". [online] World Health Organisation. Available at: apps.who.int/iris/bitstream/10665/255540/1/9789241512459-eng.pdf?ua=1 [Accessed 8 Feb. 2018].

4 See "Access to treatment: Unequal care for European rare disease patients" [online] Available at: download.eurordis.org.s3.amazonaws.com/Access%20to%20treatment/access%20to%20treatment%20EN.pdf [Accessed 8 Feb. 2018].

5 See "Estudio sobre situación de Necesidades Sociosanitarias de las personas con Enfermedades Raras en España – Estudio ENSERio" [online] Available at: www.enfermedades-raras.org/images/stories/documentos/Estudio_ENSERio.pdf [Accessed 8 Feb. 2018].

- Similarly, *availability* remains very much an aspiration and a pressing need for the global rare disease community, in a time when new treatments for an immense majority of the rare diseases known to this day are yet to be invented, when more must be done to facilitate the procurement and distribution of those medicines to those population groups and in those countries where they are needed most, or when public information about rare diseases could also be made more widely available to the general public. Recent **findings indicate that nearly a quarter of persons with rare diseases in Europe did not get the medical treatment they needed in the past year because the treatment was not available in their country**.6

- The objective of *participation* is compromised by the frequent marginalisation of persons with rare diseases as an **"invisible"** population, by the burden of caregiving duties often borne by their relatives, and by the overall discrimination and stigma still often associated with rare diseases in our societies.

- Persons with rare diseases share **the same inalienable right as all others** to life, health, *good quality* of life and holistic development and *good quality* of treatment and care.

These aspects have been amply **recognised by the UN Committee on Economic, Social and Cultural Rights (CESCR)** in its General Comment No. 14 on "*The Right to the Highest Attainable Standard of Health*"7 from which it can be directly derived that the "right to health" means that persons with rare diseases must have a sufficient availability of promotive, preventive, curative, rehabilitative and supportive/palliative healthcare services, medicines, health devices and technologies that are of a reasonable standard of quality, safety, accessibility, acceptability and affordability, and that are delivered to them without discrimination or financial ruin.

We also contend that, from a broader perspective, **the specific nature of rare diseases also calls for a holistic, comprehensive and multi-disciplinary response, deeply grounded by essence in a human rights vision**.

This very need is starting to receive wider recognition and support, as illustrated by the reflections of UN Special Rapporteur on the Right to Health Prof. Dainius Puras on the occasion of the Rare Diseases International Policy Event "*The Right to Health: The Rare Disease Perspective*" on 10 February 2017 in Geneva.8

Rare diseases must be addressed as a human rights and public health issue insofar as:

- Persons, and particularly children, affected by rare diseases have a right to life, health, good quality of life and holistic development and good quality of treatment and care

- Existing gaps in research, early identification and treatment of rare diseases need to be closed

- More must be done to encourage the research and development of needed medicines, and to facilitate the procurement and distribution of those medicines to those population groups and in those countries where they are needed most

- States and public authorities must ensure existing medicines, other non-pharmacological treatments and assistive devices are affordable and accessible on a non-discriminatory basis, and where necessary in low-cost forms or versions for low-resource countries9

- A comprehensive approach to rare diseases is needed, encompassing education, research, prevention, diagnosis, care and treatment, social support and inclusion

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6 See "Access to treatment: Unequal care for European rare disease patients" [online] Available at: download.eurordis.org.s3.amazonaws.com/Access%20to%20treatment/access%20to%20treatment%20EN.pdf [Accessed 8 Feb. 2018].

7 As adopted at the 22nd CESCR Session on 11 August 2000, and accessible here: www.refworld.org/pdfid/4538838d0.pdf

8 See "*The Right to Health and Rare Diseases*" [online] Available at: download.eurordis.org.s3.amazonaws.com/rdi/2.Puras%20Presentation-%20Rare%20diseases.pptx [Accessed 8 Feb. 2018].

9 Further to the objectives of the World Health Organisation's Global Cooperation on Assistive Technology (GATE) Project, information on which can be accessed here: www.who.int/phi/implementation/assistive_technology/phi_gate/en/

- Services and support for patients and families affected by rare diseases need to be holistic and integrated to provide for the health, disability and social issues often associated with them
- Specific funding from governments for research projects on rare diseases is needed
- International patient registries and biospecimen repositories for rare diseases should be encouraged and available for appropriate use by the rare diseases community

To conclude, and based on the preceding elements, it is our firm belief that:

- There is a pressing need for the specificities of persons with rare diseases to be better understood and recognised, at all levels, so that such minority groups cease to be subject to discrimination, neglect or exclusion.
- **States have a crucial duty to raise awareness and increase knowledge about rare diseases**, and about the profound everyday consequences faced by affected individuals. This cannot be underestimated as professionals in the areas of healthcare, social services, education and employment – on whom persons with rare diseases depend in all aspect of their everyday lives – very often do not have sufficient information to properly handle the special needs of people living with rare diseases.
- **Rare diseases offer a unique and privileged terrain for the practical application of the "right to health" framework** towards the realisation of the objectives assigned to the SDGs, but also for the definition of concrete solutions that will offer tangible metrics of progress on the path to 2030.

While the vast majority of existing initiatives and efforts to deliver the objectives of the SDGs may benefit persons with rare diseases to a certain extent, in many cases they may not because of the unique nature of these small populations whose conditions are often undiagnosed, whose symptoms are not acknowledged, and whose disease progression is undocumented. **Specific compensatory actions are required, both in the form of strategies and frameworks addressing all rare diseases, and/or in the form of distinct actions for rare diseases under and within each SDG.**

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*Rare Diseases International (RDI) and European Organisation for Rare Diseases (EURORDIS), NGOs without consultative status, also share the views expressed in this statement.